Applicants: Nathan Ellis, James German, and Joanna Groden

Serial No.: 09/753,143 Filed: January 2, 2001

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## Amendments to the Claims:

Please amend Claims 11 and 86-93 as set forth below.

## 1-10. (Canceled)

- 11. (Currently Amended) A method for determining whether a subject is a carrier of a mutated *BLM* gene comprising detecting the presence <u>or absence</u> of a mutated *BLM* gene in nucleic acid of said subject, wherein the mutated *BLM* gene is a mutated form of nucleic acid encoding the amino acid sequence set forth in SEQ ID NO:78 <u>and wherein</u> the mutated <u>BLM</u> gene is located between loci D15S1108 and D15S127 of human <u>chromosome 15</u>.
- 12. (Original) The method of Claim 11 wherein the subject is an embryo, fetus, newborn, infant or adult.
  - 13. (Canceled)
  - 14. (Original) The method of Claim 11 wherein the nucleic acid is DNA or RNA.
- 15. (Original) The method of Claim 11 wherein the presence of the mutated *BLM* gene is detected by one or more techniques selected from the group consisting of sequence analysis, restriction enzyme digestion analysis, hybridization and polymerase chain reaction.
- 16. (Original) The method of Claim 11 wherein the presence of the mutated *BLM* gene is detected by the presence of a gene product encoded by the mutated *BLM* gene.

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17. (Original) The method of Claim 16 wherein the gene product is a protein.

18. (Canceled)

19. (Original) The method of Claim 16 wherein the gene product is mRNA.

20. (Original) The method of Claim 19 wherein the mRNA is detected by one or more techniques selected from the group consisting of sequence analysis, hybridization and polymerase chain reaction.

## 21-85. (Canceled)

- 86. (Currently amended) The method of Claim 11, wherein the mutated *BLM* gene is characterized by comprises one or more of: (i) a deletion of nucleotides 631-633 of SEQ ID NO:72; (ii) a substitution of A with T at nucleotide 888 of SEQ ID NO:72; (iii) an insertion of A after nucleotide 1610 of SEQ ID NO:72; (iv) a substitution of A with G at nucleotide 2089 of SEQ ID NO:72; (v) a replacement of nucleotides ATCTGA at position 2281-2286 of SEQ ID NO:72 with nucleotides TAGATTC; (vi) a substitution of T with C at nucleotide 2596 of SEQ ID NO:72; and (vii) a substitution of G with C at nucleotide 3238 of SEQ ID NO:72.
- 87. (Currently amended) The method of Claim 86, wherein the mutated *BLM* gene comprises is characterized by a deletion of nucleotides 631-633 of SEQ ID NO:72.
- 88. (Currently amended) The method of Claim 86, wherein the mutated *BLM* gene comprises is characterized by a substitution of A with T at nucleotide 888 of SEQ ID

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NO:72.

- 89. (Currently amended) The method of Claim 86, wherein the mutated BLM gene comprises is characterized by an insertion of A after nucleotide 1610 of SEQ ID NO:72.
- 90. (Currently amended) The method of Claim 86, wherein the mutated BLM gene comprises is characterized by a substitution of A with G at nucleotide 2089 of SEQ ID NO:72.
- 91. (Currently amended) The method of Claim 86, wherein the mutated BLM gene comprises is characterized by a replacement of nucleotides ATCTGA at position 2281-2286 of SEQ ID NO:72 with nucleotides TAGATTC.
- 92. (Currently amended) The method of Claim 86, wherein the mutated BLM gene comprises is characterized by a substitution of T with C at nucleotide 2596 of SEQ ID NO:72.
- 93. (Currently amended) The method of Claim 86, wherein the mutated BLM gene comprises is characterized by a substitution of G with C at nucleotide 3238 of SEQ ID NO:72.